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Chapter 9

Summary



Training in genetics and genomics for primary health care workers

Medical professionals in primary care are increasingly expected to deliver genetic services in daily patient care and need to be prepared for patients asking for information and advice on genetics. This requires appropriate skills and knowledge of genetics needed for daily practice. However, postgraduate (physician training) and master (midwifery training) programmes in primary care and public health are currently failing to meet these perceived educational needs. Improvements in genetics education for primary care providers are thus needed to keep up with the rapid developments in genetics/genomics.

The main objectives of this study project were to reflect on current genetics/genomics developments with primary care workers, to help them identify their learning priorities and to evaluate three CPD modules in oncogenetics developed in collaboration with multidisciplinary team of general practitioners (GPs), educationalists and clinical geneticists familiar with genetics in primary care. Key factors for successful future training were identified and could make integrating genetics step by step in daily genetic primary care possible.

In the first part of this thesis an agenda for effective genetic educational strategies was created for which a needs assessment and prioritization of genetic education in primary care were studied.

Chapter 2 presents the results of a focus group study, which explored the role of genetics in primary care (i.e. family medicine and midwifery care) and the need for education in this area as perceived by primary care health workers, patient advocacy groups and clinical genetics professionals. Forty-four participants took part in three types of focus groups: mono-disciplinary groups of general practitioners and midwives, respectively and multidisciplinary groups composed of a diverse set of experts. Four themes emerged regarding the educational needs and the role of genetics in primary care: (1) the need for genetics knowledge, (2) taking a family

history, (3) ethical dilemmas and psychosocial effects in relation to genetics, and (4) insight into the organisation and role of clinical genetics services. The role of genetics in primary care was perceived to be unduly limited as a result of care providers' inadequate genetics knowledge and skills. Although all focus group participants acknowledged the importance of genetics education, general practitioners seemed to feel this need more urgently than midwives and more strongly emphasized their perceived knowledge deficiencies. The study results indicated that Dutch primary care providers need, and would welcome, more extensive education in genetics, for instance in postgraduate and master programmes.

Chapter 3 presents the results of a Delphi consensus procedure, which aimed to operationalize the focus group results and prioritise topics for genetics education for general practice. Topics mentioned as learning goals were rephrased in line with core competences. The study conducted, consisted of three rounds. A purposively selected heterogeneous panel ($n=18$) of experts, comprising six practising GPs also engaged in research, five GP trainers, four clinical genetics professionals and three representatives of patient organisations, participated. Educational needs regarding genetics in general practice in terms of knowledge, skills and attitudes, were rated and ranked in a top 10. The entire panel completed all three rounds. Kendall's coefficient of concordance indicated significant agreement regarding the top ten genetic educational needs ($P<0.001$). "Recognising signals that are potentially indicative of a hereditary component of a disease" was rated highest, followed by "Evaluating indications for referral to a clinical genetics centre" and "Knowledge of the possibilities and limitations of genetic tests". It was concluded that the education priorities resulting from the study could be used to guide the development of genetics genetic education to improve GP performance in daily practice.

The results described in Chapters 2 and 3 informed the development of genetic Continuing Professional Development (CPD) modules, including input for case-based education, to improve GP performance in genetic patient care. More research into the educational priorities in genetics is needed to design courses that are suitable for master programmes for midwives.

The second part of this thesis describes the development and evaluation of three training modules: a Genetic online Continuing Professional Development (G-eCPD) and live genetic CPD module, taking oncogenetics as an example, supported by a website on genetics for GPs (*huisartsengenetica.nl* or "*GP and genetics*"). Identification of patients at risk for hereditary cancers is considered essential to inform decisions about early screening, genetic testing, and pre-symptomatic risk reducing options. To our knowledge these were the first randomized controlled trials

(RCTs) to investigate improvement of GPs' oncogenetic knowledge and professional behavior after participation in educational modules.

In Chapter 4 an RCT was conducted to evaluate the educational outcomes of a G-eCPD module at the first two levels of the Kirkpatrick framework (satisfaction and learning). The aim of this G-eCPD module was to provide physicians sufficient knowledge and skills through didactic presentations, interactive cases and enabling tools such as information about regional possibilities for referral and consultation. It meant to enable GPs to identify patients with an inherited predisposition to cancer; draw a family tree as a tool for identifying patients at risk for hereditary cancer; describe the most common types of hereditary cancer (i.e. breast, colon) and the likely genetic mutations involved; apply oncogenetics guidelines in identifying patients for whom referral is indicated or not and find relevant information online; explain the possibilities and limitations of oncogenetic testing; discuss with patients periodic examinations and risk-reducing surgical options that are available to patients with hereditary cancer.

The G-eCPD module aimed at improving GPs' knowledge about oncogenetics, and was conducted between September 2011 and March 2012. The study method was a parallel-group pre-post-retention (six-month follow-up) controlled group intervention trial, with repeated measurements. Of the total of 80 Dutch GP volunteers (40 intervention group and 40 control group randomly assigned), 44 participants (20 intervention, 24 control group) completed all the learning activities, knowledge tests, and questionnaires. For validity reasons, recruitment was limited to all GPs practicing outside the two Dutch provinces in the North (Noord Holland) and South (Limburg) where GPs could also participate in the live CPD module. The findings of the RCT showed that satisfaction with the module was high, with the three item's scores in the range 4.1-4.3 (five-point scale) and a global score of 7.9 (ten-point scale). Knowledge gains at post-test and retention test were 0.055 ($P < 0.05$) and 0.079 ($P < 0.01$), respectively, with moderate effect sizes (0.27 and 0.31). The participants appreciated the applicability in daily practice of knowledge aspects (item scores 3.3-3.8, five-point scale), but scores on self-reported identification of disease, referral to a specialist, and knowledge about the possibilities/limitations of genetic testing were near neutral (2.7-2.8, five-point scale). It was concluded that the educational effects reported in this study can inform further development of online G-eCPD aimed at improving physicians' genetics knowledge and could potentially improve patient care. The online CPD module with its framework could reach a large group of physicians with a wide variety of backgrounds, but adapted to its audience, possibly also medical school students and be used in high school biology classes.

Chapter 5 describes whether a live oncogenetics CPD module improves GP consultation skills. In this pragmatic, blinded RCT, the intervention consisted of a four-

hour training (December 2011 and April 2012), covering oncogenetic consultation skills (family history, familial risk assessment, and efficient referral), attitude (medical ethical issues), and clinical knowledge required in primary care consultations. Outcomes were measured using observation checklists by unannounced standardized patients and self-reported questionnaires. For logistic reasons, recruitment was limited to all GPs practicing two Dutch provinces in the North (Noord Holland) and South (Limburg) who did not participate in the evaluation of the G-eCPD. Of 88 randomized GPs who initially agreed to participate, 56 completed all measurements. Key consultation skills trained equipped the GP to recall clinically relevant information about types of hereditary cancer (breast, ovarian, colon, skin) including genes associated with oncogenetics syndromes most commonly tested for; recognize patients with features suggesting inherited predisposition to cancer; draw a family tree as a tool to identify patients at risk; discuss (possible) familial and hereditary cancer risks, management of potentially developing hereditary cancer (i.e. surveillance and risk-reducing surgical options) and related ethical issues; identifying patients for referral for risk assessment and find relevant information online using oncogenetics guidelines; explain the possibilities and limitations of oncogenetic testing; and know when to consult and/or refer to a genetics specialist. These key consultation skills significantly and substantially improved; regression coefficient post-intervention equal to .34 and .28 at 3-month-follow-up indicating moderate effect size. The results show sustained improvement three months after the training as well as high satisfaction with the training and positive perceptions of the practical applicability of training topics.

Chapter 6 aimed to determine long-term (self-reported) genetic consultation skills among GPs who participated in the G-eCPD and live CPD modules (i.e. increased genetics awareness and referrals to clinical genetics centers), and interests in and satisfaction with the website. The genetics CPD modules achieved sustained improvement of oncogenetic competencies. Participants reported to be more alert of genetic problems. 88% of those who attended the live training reported to more frequently refer patients to the Clinical Genetics centers, compared to 29% of those who attended the online oncogenetics training. No significant change in referral numbers however was reported by the Clinical Genetics centers before and one year after the training. Moreover, sustained interest in and satisfaction with the newly developed GP website were investigated among website visitors who completed the pop-up questionnaire. Only a small number (38 visitors, 22 of these were GPs) completed the questionnaire during the 1- month study period. Satisfaction with the website and perceived applicability of the website on appropriateness of referrals however were highly scored. Website visitor numbers are increasing; with the page most often consulted "family tree drawing".

Thus, self-perceived genetic consultation skills increase long-term and there is interest in and satisfaction with the supportive website.

In Chapter 7 a step-by-step roadmap was proposed to integrate genetics in the Electronic Patient Record (EPR) in Family Medicine and clinical research. This could make urgent operationalization of readily available genetic knowledge feasible in clinical research and consequently improved medical care. Improving genomic literacy by training and education is needed first. The second step is the improvement of the possibilities to register the family history in such a way that queries can identify patients at risk. Adding codes to the EPR in the International Classification of Primary Care (ICPC) in the ICPC chapters “A21 Personal/family history of malignancy” and “A99 Disease carrier not described further” and other coding strategies for simple registry of family history and develop and support coding skills is proposed. EPRs need possibilities to add (new) family history information, including links between individuals who are family members. Multidisciplinary guidelines for referral must be unambiguous. Automatic alerts should help GPs to recognize patients at risk who satisfy referral criteria. A familial breast cancer case with a *BRCA1* mutation as an example was used for illustration.

Concluding remarks

The results of the studies presented in this thesis have provided better insight into how non-genetic specialists (e.g. GPs and midwives) perceive the increased role for genetics in primary care and consequently recognized the importance of genetics education. The responsibilities of primary care providers with regard to genetics require further study. The GP-specific oncogenetic CPD modules supported by the Dutch genetics website *huisartsengenetica.nl*, suggest to be a feasible, satisfactory and clinically applicable method to improve oncogenetics competences in daily practice and suggests to be an adaptable and effective educational framework to inform future training activities with the ultimate aim of improving genetic medical care.